

# **NIH Office of Rare Diseases Current and Future Activities States as Part of a Global Approach**

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# Office of Rare Diseases (ORD) -

## Collaborative Programs

- **Intramural Research and Training Programs**
  - **Bench to Bedside Research Program with Extramural Research Programs**
  - **Clinical and Biochemical Genetics Training Program (NHGRI/ Clinical Center)**
  - **OTT/ORD Technology Transfer Activities of Discoveries**
  - **Unknown Diagnosis (Diseases of Unknown Origin)**
  
- **Extramural Research Program**
  - **Scientific Conferences > 845 To identify research opportunities and to develop a research agenda for a specific disease(s)**
  - **Rare Diseases Clinical Research Network (RDCRN): Re-competition (2008-2009)**
  - **CETT Genetic Testing Program – Information and Standard Report Forms**

# Office of Rare Diseases (ORD) - Collaborative Programs

- **Inventory of Bio-specimen Collection, Storage, and Distribution Systems (RAND Corporation) (Euro Bio Bank) > 30 million specimens have been collected specifically for research purposes and Stored at >300 bio-specimen repositories in the United States**
- **Conference on Research Resources, Chemical Libraries and Screening Programs Available for Rare Diseases (Planning stages – Volunteers needed)**
- **National Coalition for Health Professional Education in Genetics (NCHPEG)**
- **Genetic and Rare Diseases Information Center**
- **Trans-NIH Working Group on Rare Diseases Research**

# **Office of Rare Diseases (ORD) - Collaborative Clinical Research Programs**

- **Angel Flight/Mercy Medical Airlift – Patient Travel**
- **Educational Program Module on Rare Diseases for Children with Office of Science Education/NIH – Global Partners Needed to Shape Content**
- **Knowledge Management for Disease - Coding of Research Projects Supported by NIH ~5000 Research Projects and >1700 Rare Diseases or Conditions ( Next Step Rare Cancers)**

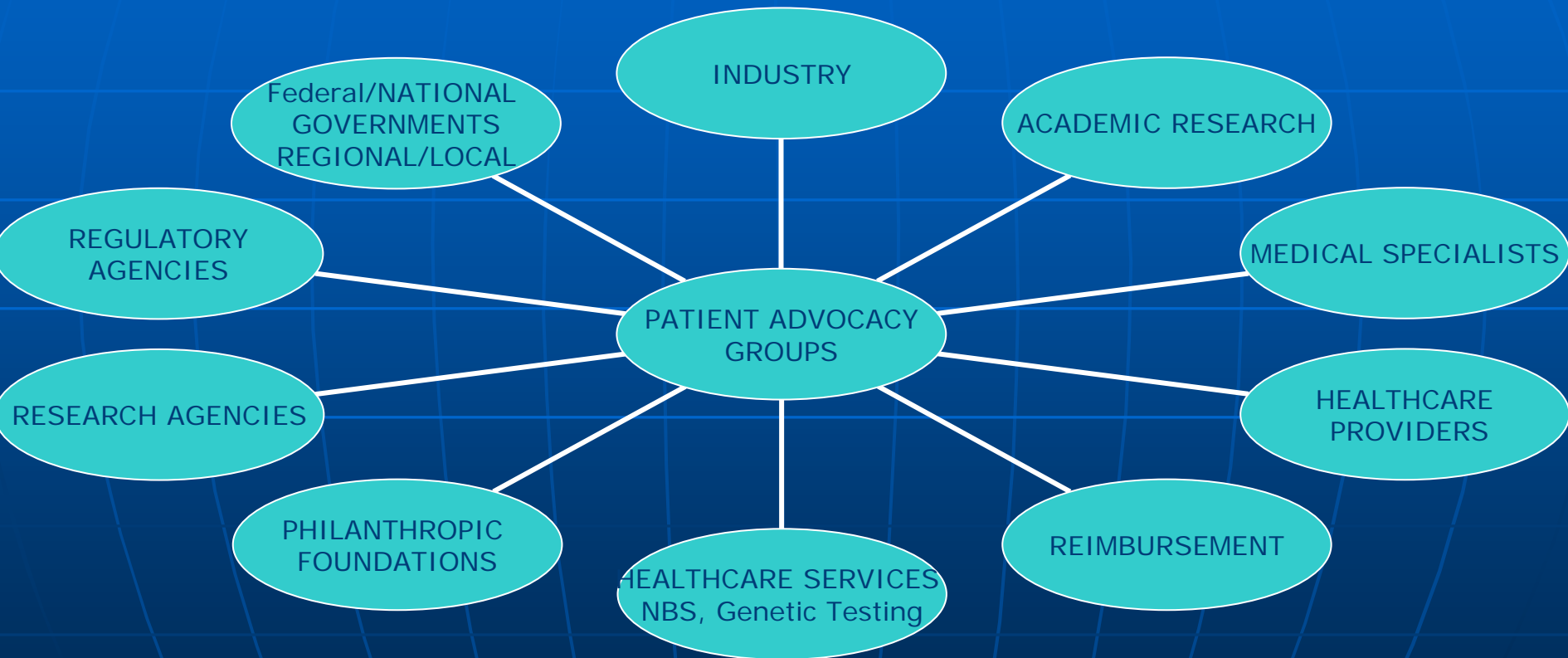
# Awaiting Advances from Current and Future Research Activities

- Genotype-Phenotype Association Studies
- Personalized Medicine - Genetics and Genomics in Medicine
- Genetic Testing Before Treatment to Identify Responders and Potential Toxicities
- Gene Therapy, Stem Cell Therapy, and Nanotechnology Advances
- Traditional Small Molecule Development Continue with Biological Learning

# Rare Diseases – Perceptions

- Very Few People Have the Condition
- Little or No Information Available
- Little or No Research Interest
- No Treatments are Available
- Limited Access to Rare Diseases-Focused Clinicians
- Little or No Hope for the Future

# One View of the Rare Diseases Community



# Current NIH Support of Drug Development for Rare Diseases (Source: Dr. Chris Austin – NHGRI)

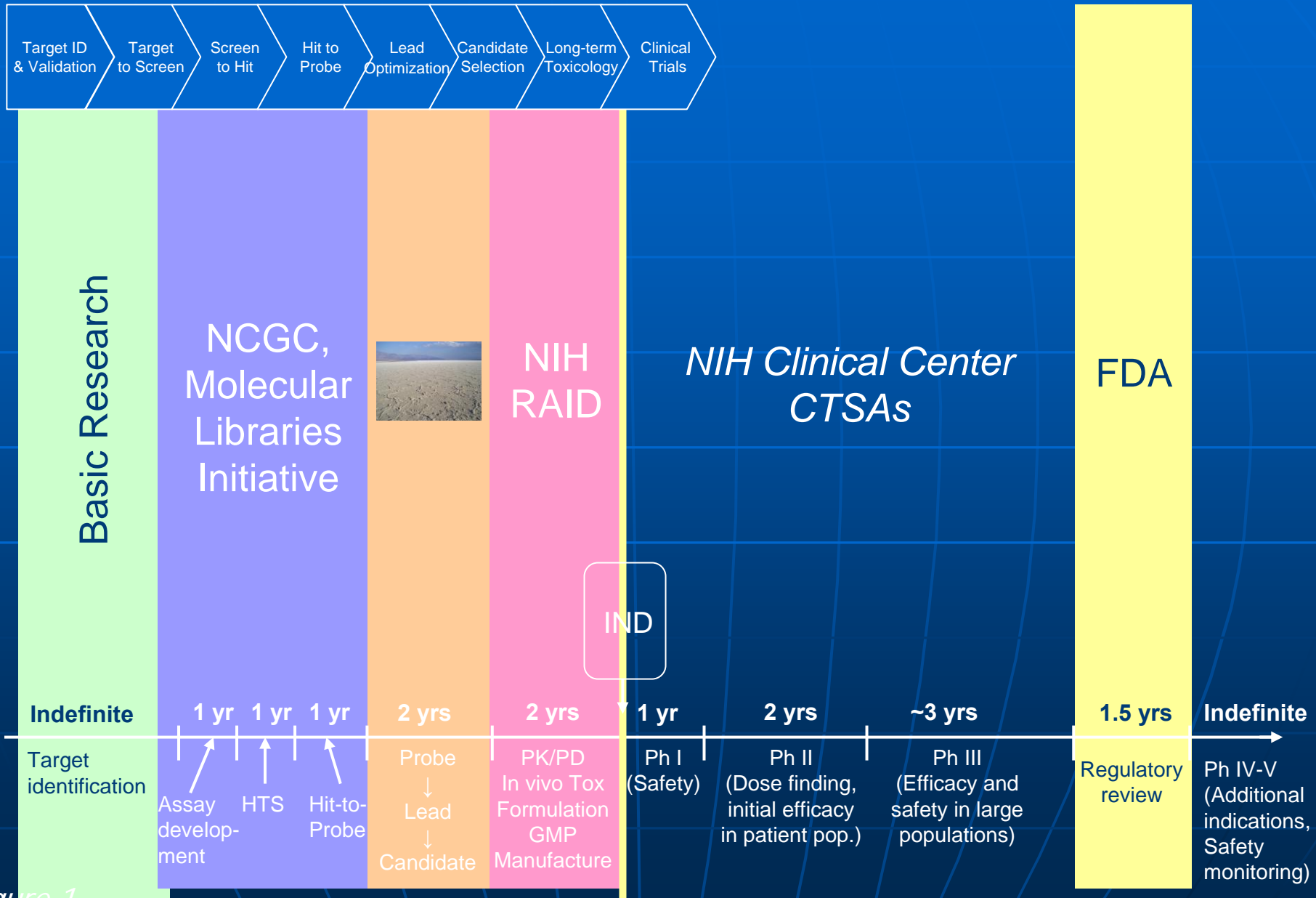


Figure 1



# Meeting the Needs of the Rare Diseases Community

- **Identify and Expand Worldwide Partnerships and Collaborations of Patient Advocacy Groups – NORD, EURORDIS, Genetic Alliance. KORD, NZORD, CORD**
- **Increase Emphasis on Rare Disease Research and Orphan Products Development Activities at National and International Levels**
- **Identify Novel Approaches to Provide Support for Research - Italy, USA SBIR/STTR Reauthorization Proposed Legislation**
- **Increased Collaborations of Research Investigators and Develop Globalization of Research Efforts with Common Protocols and Multidisciplinary Research Teams (Treat NMD, Prader-Willi Syndrome, Progeria)**
  - **Industry**
  - **Academia**
  - **Government**
  - **Foundations**

# Meeting the Needs of the Rare Diseases Community

- **Continue Collaborative Efforts for Acceptance of Research Data for Regulatory Purposes**
  - Gain Acceptance of Study Design and Bio-statistical Analyses for Small Patient Sample Sizes
- **Establish Better Definitions of Patient Responders with Development of Appropriate Biomarkers and Surrogate Endpoints for Safety and Efficacy/Genetic Predictors of Responders**
- **Link Information Resources**
  - Clinical Studies at Multi-National Research Sites (Phase 1-4, Longitudinal, Natural History, Observational)
  - Treatment Sites
  - Genetic Tests and Materials Standards
  - Rare Diseases Information
  - Patient Advocacy Groups
- **Increase Awareness and Information Level About Rare Diseases and Orphan Products with educational programs for...**
  - Health Care Providers
  - Patients
  - Public
  - Media

# Meeting the Needs of the Rare Diseases Community

- **Improve Health Literacy of Populations to Enable Information-Based Decision Making on Living and Coping with Rare and Genetic Diseases**
- **Develop Better Tools for Patient Recruitment and Referral (Internet Based)**
- **Coordinate Utilization and Access to Public and Private Databases and Results of ...**
  - **High Throughput Screening Processes**
  - **Chemical Genomics Screening and Re-Purposing of Approved and Investigational Products**
    - **Continue Small Molecule Discoveries/Medicinal Chemistry Applications**
  - **Develop Procedures to Provide Access to Molecular Libraries and Databases**
- **Resolve Intellectual Property Issues that are Barriers to R & D**

# Information Development, Dissemination, and Education Activities – ORD Website

- National Library of Medicine Gateway  
<http://gateway.nlm.nih.gov/gw/Command>
- Research Projects - CRISP  
<http://crisp.cit.nih.gov/>
- Patient Advocacy Groups - NORD, Genetic Alliance, DURLINE, and EURORDIS > 1200
- Clinical Trials.gov (Total >55,100 studies from 155 Countries, and 50,000 users/day
  - >7,400 Studies Recruiting for 1,100 Rare Diseases (>16,400 Total for >1,240 Rare Diseases)
- Pub Med/MEDLINE – 4800 Journals from 70 Countries, 750 Million Searches/year
- Gene Tests (1549 Diseases) - 1264 Clinical Laboratories - 285 Diseases Research Only Laboratories

# Office of Rare Diseases – Extended Staff

- Ms. Mary Demory
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- David Eckstein, Ph.D.
- John Ferguson, M.D.
- Rashmi Gopal-Srivastava, Ph.D.
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